

APPROACH TO CHILD IMMUNODEFICIENCY



INTRODUCTION

- SYSTEMS INVOLVED IN IMMUNODEFICIENCY:

1-the adaptive immune system:

humoral

cellular

2-the innate immune system:

phagocytes

complement

- **Primary immunodeficiency is an inherited defect in these systems.**

- **EPIDEMIOLOGY**

Overall, about 1:500 are born with and 1:1200 currently live with a PID.

SECONDARY IMMUNODEFICIENCY

Causes: acquired (not genetic) as

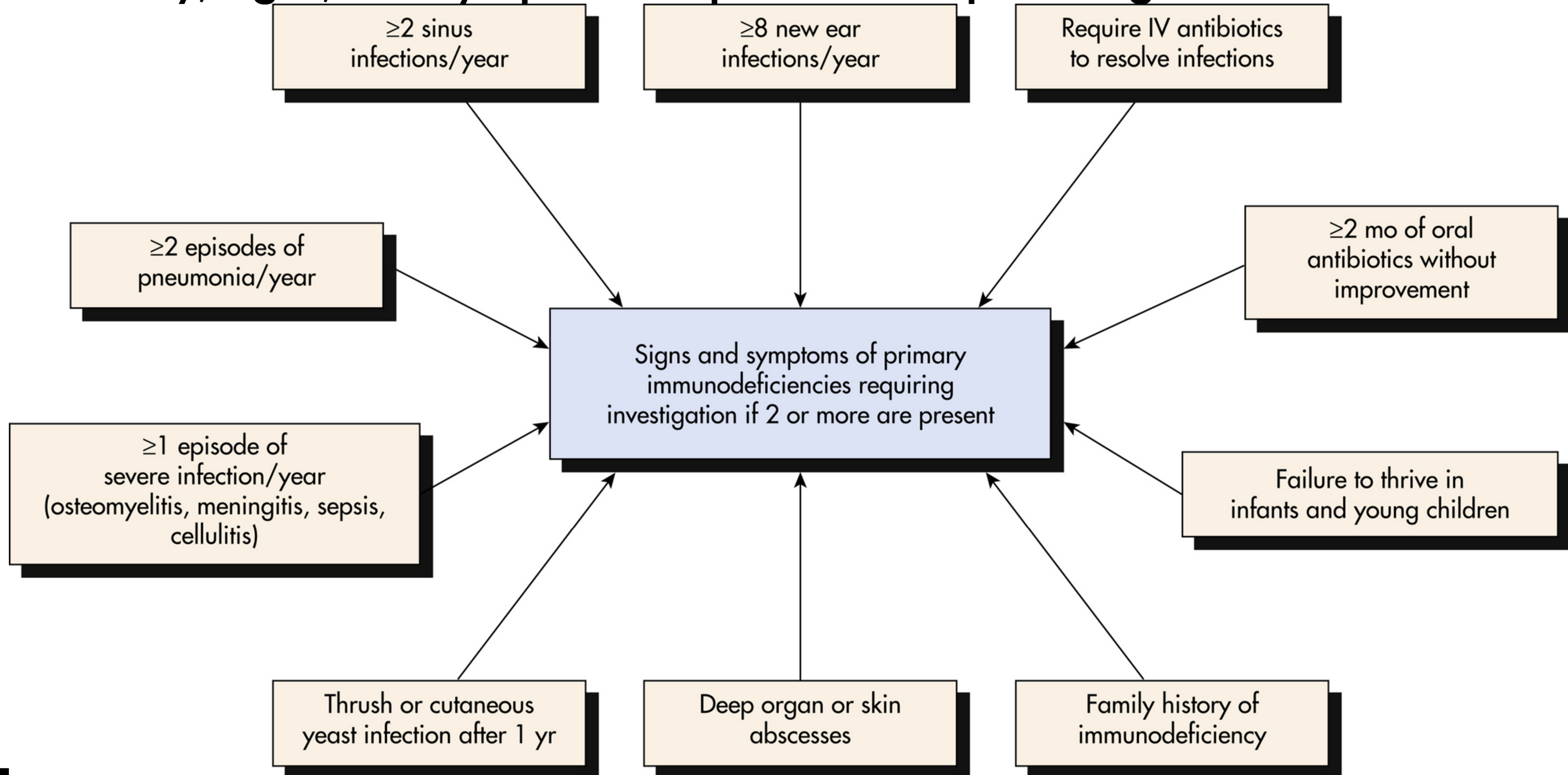
- Infections: HIV
- Anatomical: Eustachian tube obstruction, lung sequestration
- Environmental: steroid therapy, diabetes mellitus, cancer, asplenia, sickle cell anemia, MRSA
- Malnutrition

COMBINED IMMUNODEFICIENCY

- SCID
- Wiskott-Aldrich syndrome

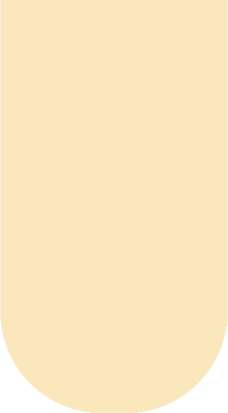
PHYSICAL FINDINGS & CLINICAL PRESENTATION

- history, signs, and symptoms depend on the precise genetic disorder



ONSET WHEN RECURRENT INFECTIONS BEGAN IS IMPORTANT

- before age 6 months suggests a T-cell defect (intracellular pathogens).
- between 6 and 12 months may suggest combined B- and T-cell defects or a B-cell defect (maternal antibodies are disappearing)
- After 12 months usually suggests a B-cell defect or secondary immunodeficiency.



CLINICAL CHARACTERISTICS OF PRIMARY IMMUNODEFICIENCIES



B-CELL DEFECTS

- Recurrent pyogenic infections with extracellular encapsulated organisms (*Streptococcus pneumoniae*, *Haemophilus influenzae* type b, and group A *Streptococcus*), *Mycoplasma* sp., and *Ureaplasma* sp., *Giardia* and enteroviruses
- Otitis, sinusitis, recurrent pneumonia, bronchiectasis, and conjunctivitis
- Few problems with fungal or viral infections (except enterovirus and poliomyelitis)
- Diarrhea common, especially secondary to infection with *Giardia lamblia* and due to enteropathy
- Minimal growth retardation

T-CELL DEFECTS

- susceptible to intracellular pathogens, including viruses(herpes simplex,cytomegalo virus), fungi (candida,pneumomyocystis jirovecii) and protozoa (toxoplasma gondii)
- Growth retardation, malabsorption, diarrhea, and failure to thrive is common
- Anergy
- Increased incidence of malignancy
- Poor survival beyond infancy or early childhood in the absence of stem cell transplantation

NEUTROPHIL DEFECTS

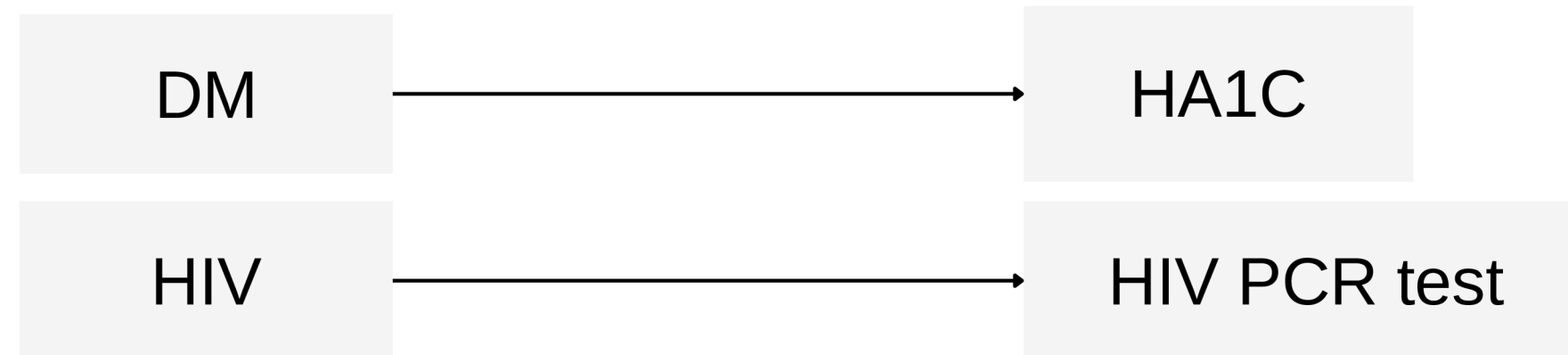
- Recurrent dermatologic infections with bacteria such as *Staphylococcus*, *Pseudomonas*, and *Escherichia coli*, *Nocardia*, and fungi such as *Aspergillus*
- Subcutaneous, lymph node, lung, and liver abscesses
- Pulmonary infections common, including abscess and pneumatocele formation, contributing to chronic disease
- Bone and joint infection common
- Delayed separation of umbilical cord
- Poor wound healing

COMPLEMENT DEFECTS

- Recurrent bacterial infections with extracellular encapsulated organisms, such as *S. pneumoniae* and *H. influenzae*
- Susceptibility to recurrent infections with *Neisseria meningitidis*
- Increased incidence of autoimmune disease
- Severe or recurrent skin and respiratory tract infection

INITIAL TESTING

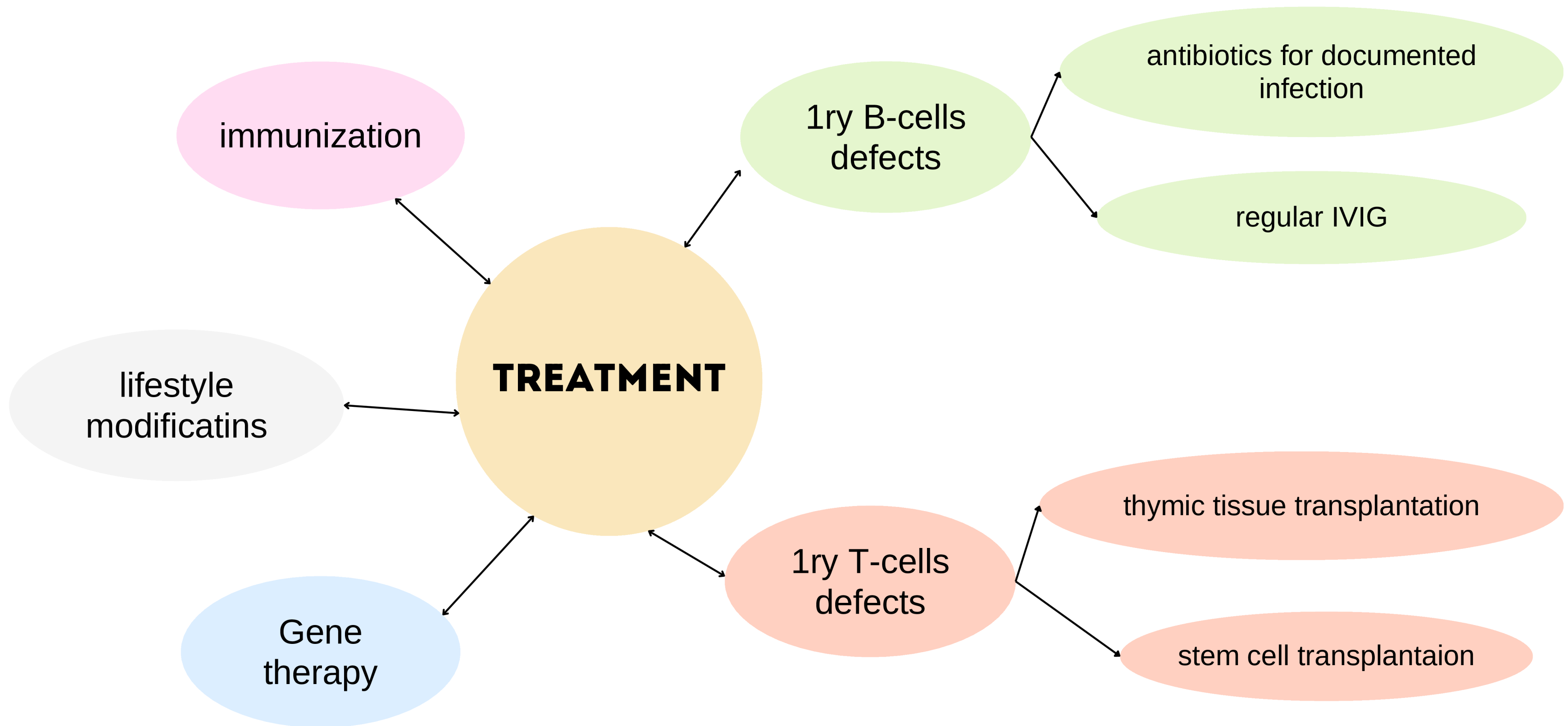
- If a specific secondary immunodeficiency disorder is suspected clinically, testing should focus on that disorder. for example:



- Initial screening tests for 1ry ID are:
 1. CBC with manual differentiation
 2. Quantitative ig measurements
 3. Antibody titers (not after blood transfusion/FFP/IVIG)
 4. Skin testing for delayed hypersensitivity.

ADVANCED INVESTIGATIONS

- **Flow cytometry as in CD3,CD4,CD8 for cellular immunity defect, and CD19,20 for humeral immunity defects**
- **functional assays: Mitogen/PHA proliferation assay (T cell function)**
- **(DHR)Dihydrorhodamine test for phagocytes defect**
- **for complement Defects CH50/AH50 Or for specific elements as C3/C4**
- **autoantibodies screen**
- **genetic test in some cases**





THANK YOU!